

I. AMENDMENT

Please cancel claims 5, 12 to 15, 38, 64, 67, 70 and 71 without prejudice, and amend the claims as indicated. Upon entry of the present amendment, the status of the claims will be as follows:

1. (Currently amended) A ~~primer~~ set of primers, comprising primers that selectively hybridize under highly stringent conditions to a nucleotide sequence flanking and within fifty nucleotides of each of polycystic kidney disease-associated protein-1 (PKD1) gene sequences set forth as

nucleotides 2043 to 4290 of SEQ ID NO:1,

nucleotides 17907 to 22489 of SEQ ID NO:1,

nucleotides 22218 to 26363 of SEQ ID NO:1,

nucleotides 26246 to 30615 of SEQ ID NO:1,

nucleotides 30606 to 33957 of SEQ ID NO:1,

nucleotides 36819 to 37140 of SEQ ID NO:1

nucleotides 37329 to 41258 of SEQ ID NO:1, and

nucleotides 41508 to 47320 of SEQ ID NO:1,

or to a nucleotide sequence complementary to said PKD1 gene sequences,

wherein a primer of said set hybridizes to a nucleotide sequence flanking and within fifty nucleotides of one of said PKD1 gene sequences,

each of said primers comprising a 5' region and adjacent 3' region,

said 5' region comprising a nucleotide sequence that selectively hybridizes to a ~~polycystic kidney disease-associated protein-1 (PKD1)~~ PKD1 gene sequence and, optionally, to a PKD1 gene homolog sequence, wherein said 5' region comprises at least ten contiguous nucleotides, and

said 3' region comprising a nucleotide sequence that selectively hybridizes to a PKD1 gene sequence, and not to a PKD1 gene homolog sequence, provided the primer does not consist of a sequence as set forth in SEQ ID NO:11, SEQ ID NO:18, SEQ ID NO:52, or SEQ ID NO:60.

2. (Currently amended) The ~~primer~~ set of primers of claim 1, wherein the 3' region comprises at least one 3' terminal nucleotide identical to a nucleotide that is 5' and adjacent to the nucleotide sequence of the PKD1 gene to which the 5' region of the primer can hybridize, and wherein said 3' terminal nucleotide is different from a nucleotide that is 5' and adjacent to a nucleotide sequence of the PKD1 homolog to which the 5' region of the primer can hybridize.
3. (Currently amended) The ~~primer~~ set of primers of claim 2, wherein the 3' region comprises ~~about~~ 2 to 4 3' terminal nucleotides.
4. (Currently amended) The ~~primer~~ set of primers of claim 2, comprising a 5' region of ~~about~~ 14 to 18 nucleotides and a 3' region of ~~about~~ 2 to 6 nucleotides.
5. (Cancelled).
6. (Currently amended) The ~~primer of claim 5~~ set of primers of claim 1, comprising a nucleotide sequence substantially identical to any of SEQ ID NOS:3 to 51 and 61 to 113.
7. (Currently amended) A set of primer pairs, each primer pair comprising a primer of claim 1, which wherein the primer pairs selectively hybridize to SEQ ID NO:1 and can amplify a portion of SEQ ID NO:1 comprising about nucleotides 2043 to 4290; nucleotides 17907 to

22489; nucleotides 22218 to 26363; nucleotides 26246 to 30615; nucleotides 30606 to 33957; nucleotides 36819 to 37140; nucleotides 37329 to 41258; nucleotides 41508 to 47320; or a combination thereof.

8. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 7, comprising a forward primer and a reverse primer, each of which is selected from SEQ ID NOS:3 to 18.

9. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 7, wherein the primer pair comprises SEQ ID NOS:3 and 4; SEQ ID NOS:5 and 6; SEQ ID NOS:7 and 8; SEQ ID NOS:9 and 10; SEQ ID NOS:11 and 12; SEQ ID NOS:13 and 14; SEQ ID NOS:15 and 16; or SEQ ID NOS:17 and 18.

10. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 7, comprising a forward primer and a reverse primer, each of which is selected from SEQ ID NOS:19 to 51 and 61 to 113.

11. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 10, wherein the primer pair comprises SEQ ID NOS:19 and 20; SEQ ID NOS:21 and 22; SEQ ID NOS:23 and 24; SEQ ID NOS:25 and 26; SEQ ID NOS:27 and 28; SEQ ID NOS:29 and 30; SEQ ID NOS:31 and 32; SEQ ID NOS:33 and 34; SEQ ID NOS:35 and 36; SEQ ID NOS:37 and 38; SEQ ID NOS:39 and 40; SEQ ID NOS:41 and 42; SEQ ID NOS:43 and 44; SEQ ID NOS:45 and 46; SEQ ID NOS:47 and 48; SEQ ID NOS:49 and 50; SEQ ID NOS: 51 and 61; SEQ ID NOS:62 and 63; SEQ ID NOS:64 and 65; SEQ ID NOS:66 and 67; SEQ ID NOS:68 and 69; SEQ ID NOS:70 and 71; SEQ ID NOS:72 and 73; SEQ ID NOS:74 and 75; SEQ ID NOS:76 and 77; SEQ ID NOS:78 and 79; SEQ ID NOS:80 and 81; SEQ ID NOS:82 and 83; SEQ ID NOS:84 and 85; SEQ ID NOS:86 and 87; SEQ ID NOS:88 and 89; SEQ ID NOS:90 and 91; SEQ ID NOS:92 and 93; SEQ

ID NOS:94 and 95; SEQ ID NOS:96 and 113; SEQ ID NOS:97 and 98; SEQ ID NOS:99 and 100;
SEQ ID NOS:101 and 102; SEQ ID NOS:103 and 104; SEQ ID NOS: 105 and 106; SEQ ID
NOS:107 and 108; SEQ ID NOS:109 and 110; or SEQ ID NOS:111 and 112.

12 to 15. (Cancelled)

16. (Currently amended) A solid matrix, comprising the ~~primer~~ the set of primers of
claim 1 ~~claim 5~~, wherein each of the primers ~~the primer~~ is immobilized on the solid matrix.

17. (Original) The solid matrix of claim 16, which comprises a plurality of immobilized
primers.

18. (Withdrawn) The solid matrix of claim 17, wherein the matrix comprises a plurality
of primers, wherein said primers are degenerate with respect to one or more codons encoding a
polypeptide having an amino acid sequence as set forth in SEQ ID NO:2.

19. (Original) The solid matrix of claim 16, wherein the solid matrix is a microchip.

20. (Previously presented) An isolated polynucleotide, comprising a contiguous sequence
of at least ten nucleotides substantially identical to a nucleotide sequence of SEQ ID NO:1 or to a
nucleotide sequence complementary thereto, the contiguous nucleotide sequence comprising with
respect to SEQ ID NO:1:

nucleotide 474, wherein nucleotide 474 is a T;

nucleotide 487, wherein nucleotide 487 is an A;

nucleotide 3110, wherein nucleotide 3110 is a C;

a position corresponding to nucleotide 3336, wherein nucleotide 3336 is deleted;

nucleotide 3707, wherein nucleotide 3707 is an A;
nucleotide 4168, wherein nucleotide 4168 is a T;
nucleotide 4885, wherein nucleotide 4885 is an A;
nucleotide 5168, wherein nucleotide 5168 is a T;
nucleotide 6058, wherein nucleotide 6058 is a T;
nucleotide 6078, wherein nucleotide 6078 is an A;
nucleotide 6089, wherein nucleotide 6089 is a T;
nucleotide 6195, wherein nucleotide 6195 is an A;
nucleotide 6326, wherein nucleotide 6326 is a T;
a position corresponding to nucleotides 7205 to 7211, wherein nucleotides 7205 to 7211 are
deleted;

nucleotide 7376, wherein nucleotide 7376 is a C;
a nucleotide sequence corresponding to nucleotides 7535 to 7536, wherein a GCG
nucleotide sequence is inserted between nucleotides 7535 and 7536;

nucleotide 7415, wherein nucleotide 7415 is a T;
nucleotide 7433, wherein nucleotide 7433 is a T;
nucleotide 7696, wherein nucleotide 7696 is a T;
nucleotide 7883, wherein nucleotide 7883 is a T;
nucleotide 8021, wherein nucleotide 8021 is an A;
a nucleotide sequence corresponding to nucleotide 8159 to 8160, wherein nucleotides 8159
to 8160 are deleted;

nucleotide 8298, wherein nucleotide 8298 is a G;
nucleotide 9164, wherein nucleotide 9164 is a G;
nucleotide 9213, wherein nucleotide 9213 is an A;
nucleotide 9326, wherein nucleotide 9326 is a T;
nucleotide 9367, wherein nucleotide 9367 is a T;

nucleotide 10064, wherein nucleotide 10064 is an A;
nucleotide 10143, wherein nucleotide 10143 is a G;
nucleotide 10234, wherein nucleotide 10234 is a C;
nucleotide 10255, wherein nucleotide 10255 is a T;
or a combination thereof.

21. (Original) A vector, comprising the polynucleotide of claim 20.

22. (Original) A host cell containing the vector of claim 20.

23. (Original) A solid matrix, comprising the polynucleotide of claim 20, wherein said polynucleotide is immobilized on the solid matrix.

24. (Original) The solid matrix of claim 23, wherein the polynucleotide comprises one of a plurality of polynucleotides, each of which is immobilized on the solid matrix.

25. (Currently amended) A method of detecting the presence or absence of a mutation in a PKD1 polynucleotide in a sample, the method comprising:

contacting nucleic acid molecules in a sample with ~~at least one primer pair~~ the set of primer pairs of claim 7 under conditions suitable for amplification of a PKD1 polynucleotide by the primer pairs ~~primer pair~~, thereby generating a PKD1-specific amplification product under said conditions; and

identifying the presence or absence of a mutation in the PKD1-specific amplification product, thereby detecting the presence or absence of a mutation in the PKD1 polynucleotide in the sample.

26. (Currently amended) The method of claim 25, wherein the ~~primer pair~~ set of primer pairs comprises SEQ ID NO:3 and 4; SEQ ID NOS:5 and 6; SEQ ID NOS:7 and 8; SEQ ID NOS:9 and 10; SEQ ID NOS:11 and 12; SEQ ID NOS:13 and 14; SEQ ID NOS:15 and 16; and SEQ ID NOS:17 and 18; ~~or a combination thereof.~~

27. (Original) The method of claim 25, wherein, upon generating a PKD1-specific amplification product, the method further comprises:

contacting the PKD1-specific amplification product with at least a second primer pair selected from SEQ ID NOS:19 and 20; SEQ ID NOS:21 and 22; SEQ ID NOS:23 and 24; SEQ ID NOS:25 and 26; SEQ ID NOS:27 and 28; SEQ ID NOS:29 and 30; SEQ ID NOS:31 and 32; SEQ ID NOS:33 and 34; SEQ ID NOS:35 and 36; SEQ ID NOS:37 and 38; SEQ ID NOS:39 and 40; SEQ ID NOS:41 and 42; SEQ ID NOS:43 and 44; SEQ ID NOS:45 and 46; SEQ ID NOS:47 and 48; SEQ ID NOS:49 and 50; SEQ ID NOS:51 and 61; SEQ ID NOS:62 and 63; SEQ ID NOS:64 and 65; SEQ ID NOS:66 and 67; SEQ ID NOS:68 and 69; SEQ ID NOS:70 and 71; SEQ ID NOS:72 and 73; SEQ ID NOS:74 and 75; SEQ ID NOS:76 and 77; SEQ ID NOS:78 and 79; SEQ ID NOS:80 and 81; SEQ ID NOS:82 and 83; SEQ ID NOS:84 and 85; SEQ ID NOS:86 and 87; SEQ ID NOS:88 and 89; SEQ ID NOS:90 and 91; SEQ ID NOS:92 and 93; SEQ ID NOS:94 and 95; SEQ ID NOS:96 and 113; SEQ ID NOS:97 and 98; SEQ ID NOS:99 and 100; SEQ ID NOS:101 and 102; SEQ ID NOS:103 and 104; SEQ ID NOS: 105 and 106; SEQ ID NOS:107 and 108; SEQ ID NOS:109 and 110; SEQ ID NOS:111 and 112; and a combination thereof, under conditions suitable for nested amplification of the PKD1-specific amplification product by the second primer pair, thereby generating a nested amplification product; and

identifying the presence or absence of a mutation in the nested amplification product, thereby detecting the presence or absence of a mutation in the PKD1 polynucleotide in the sample.

28. (Original) The method of claim 25, wherein amplification is performed by a polymerase chain reaction.

29. (Original) The method of claim 25, wherein the PKD1 polynucleotide is a variant PKD1 polynucleotide.

30. (Withdrawn) The method of claim 29, wherein the variant PKD1 polynucleotide comprises a nucleotide sequence substantially identical to SEQ ID NO:1, wherein nucleotide 474 is a T; nucleotide 487 is an A; nucleotide 4884 is an A; nucleotide 6058 is a T; nucleotide 6195 is an A; nucleotide 7376 is a C; nucleotide 7696 is a T; nucleotide 8021 is an A; nucleotide 9367 is a T; nucleotide 10143 is a G; nucleotide 10234 is a C; or nucleotide 10255 is a T.

31. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product comprises determining the nucleotide sequence of the amplification product.

32. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product comprises determining the melting temperature of the amplification product, and comparing the melting temperature to the melting temperature of a corresponding nucleotide sequence of SEQ ID NO:1.

33. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product is performed using denaturing high performance liquid chromatography.

34. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product is performed using matrix-assisted laser desorption time of flight mass spectrometry.

35. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product is performed using high throughput conformation-sensitive gel electrophoresis.

36. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product is performed by a method selected from single stranded conformation analysis, denaturing gradient gel electrophoresis, an RNase protection assay, allele-specific oligonucleotide detection, an allele-specific polymerase chain reaction, and an oligonucleotide ligation assay.

37. (Original) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product is performed using a primer extension reaction assay,
wherein the primer extension reaction is performed using a detectably labeled primer and a mixture of deoxynucleotides and dideoxynucleotides, and
wherein the primer and mixture are selected so as to enable differential extension of the primer in the presence of a wild type PKD1 polynucleotide as compared to a mutant PKD1 polynucleotide.

38. (Cancelled)

39. (Original) The method of claim 25, wherein the method is performed in a high throughput format using a plurality of samples.

40. (Original) The method of claim 39, wherein plurality of samples are in an array.
41. (Original) The method of claim 40, wherein the array comprises a microtiter plate.
42. (Original) The method of claim 40, wherein the array is on a microchip.
43. (Currently amended) The method of claim 25, wherein identifying the presence or absence of a mutation in the amplification product comprises:
- contacting the amplification product with the polynucleotide of claim 20, under highly stringent conditions ~~condition~~ suitable for selective hybridization of the polynucleotide to an identical nucleotide sequence; and
 - detecting the presence or absence of selective hybridization of the polynucleotide to the amplification product,
 - wherein the detecting the presence of selective hybridization identifies the presence of a mutation in the PKD1 polynucleotide in the sample, and
 - wherein detecting the absence of selective hybridization identifies the absence of a mutation in the PKD1 polynucleotide in the sample.
44. (Currently amended) A method of identifying a subject at risk for a PKD1-associated disorder, the method comprising:
- contacting nucleic acid molecules in a sample from a subject with ~~at least one primer pair~~ the set of primer pairs of claim 7 under conditions suitable for amplification of a PKD1 polynucleotide by the primer pair, thereby generating an amplification product; and
 - detecting the presence or absence of a mutation indicative of a PKD1-associated disorder in the amplification product,

wherein the absence of the mutation identifies the subject a not at risk for a PKD1-associated disorder, and

wherein the presence of the mutation identifies the subject as at risk for a PKD1-associated disorder.

45. (Currently amended) The method of claim 44, wherein the ~~at least one primer pair is selected from~~ set of primer pairs comprises SEQ ID NO:3 and 4; SEQ ID NO:5 and 6; SEQ ID NOS:7 and 8; SEQ ID NOS:9 and 10; SEQ ID NOS:11 and 12; SEQ ID NOS:13 and 14; SEQ ID NOS:15 and 16; and SEQ ID NOS:17 and 18.

46. (Original) The method of claim 44, wherein the PKD1-associated disorder is autosomal dominant polycystic kidney disease.

47. (Original) The method of claim 44, wherein the PKD1-associated disorder is acquired cystic disease.

48. (Original) The method of claim 44, wherein the method is performed in a high throughput format.

49. (Original) The method of claim 44, wherein detecting the presence or absence of a mutation indicative of a PKD1-associated disorder in the amplification product comprises accumulating data representative of the presence or absence of the mutation.

50. (Original) The method of claim 49, wherein the data is formatted into a report indicating whether a subject is at risk of a PKD1-associate disorder.

51. (Original) The method of claim 50, further comprising transmitting the report to a user.

52. (Original) The method of claim 51, wherein transmitting the report comprises sending the report over the internet, by facsimile or by mail.

53. (Original) The method of claim 44, further comprising contacting the amplification product with at least a second primer pair, under conditions suitable for nested amplification of the amplification product by a second primer pair, thereby generating a nested amplification product,

and detecting the presence or absence of a mutation indicative of a PKD1-associated disorder in the nested amplification product.

54. (Original) The method of claim 53, wherein the second primer pair is selected from SEQ ID NOS:19 and 20; SEQ ID NOS:21 and 22; SEQ ID NOS:23 and 24; SEQ ID NOS:25 and 26; SEQ ID NOS:27 and 28; SEQ ID NOS:29 and 30; SEQ ID NOS:31 and 32; SEQ ID NOS:33 and 34; SEQ ID NOS:35 and 36; SEQ ID NOS:37 and 38; SEQ ID NOS:39 and 40; SEQ ID NOS:41 and 42; SEQ ID NOS:43 and 44; SEQ ID NOS:45 and 46; SEQ ID NOS:47 and 48; SEQ ID NOS:49 and 50; SEQ ID NOS:51 and 61; SEQ ID NOS:62 and 63; SEQ ID NOS:64 and 65; SEQ ID NOS:66 and 67; SEQ ID NOS:68 and 69; SEQ ID NOS:70 and 71; SEQ ID NOS:72 and 73; SEQ ID NOS:74 and 75; SEQ ID NOS:76 and 77; SEQ ID NOS:78 and 79; SEQ ID NOS:80 and 81; SEQ ID NOS:82 and 83; SEQ ID NOS:84 and 85; SEQ ID NOS:86 and 87; SEQ ID NOS:88 and 89; SEQ ID NOS:90 and 91; SEQ ID NOS:92 and 93; SEQ ID NOS:94 and 95; SEQ ID NOS:96 and 113; SEQ ID NOS:97 and 98; SEQ ID NOS:99 and 100; SEQ ID NOS:101

and 102; SEQ ID NOS:103 and 104; SEQ ID NOS: 105 and 106; SEQ ID NOS:107 and 108; SEQ ID NOS:109 and 110; SEQ ID NOS:111 and 112; and a combination thereof.

55. (Original) The method of claim 53, detecting the presence or absence of the mutation comprises determining the nucleotide sequence of the amplification product, and comparing the nucleotide sequence to a corresponding nucleotide sequence of SEQ ID NO:1.

56. (Original) The method of claim 53, wherein detecting the presence or absence of the mutation comprises determining the melting temperature of the amplification product, and comparing the melting temperature to the melting temperature of a corresponding nucleotide sequence of SEQ ID NO:1.

57. (Original) The method of claim 53, wherein detecting the presence or absence of the mutation is performed using denaturing high performance liquid chromatography.

58. (Withdrawn) The method of claim 44, wherein the mutation indicative of a of PKD1 associated disorder comprises a nucleotide sequence substantially identical to SEQ ID NO:1, wherein nucleotide 3110 is a C; nucleotide 8298 is a G; nucleotide 9164 is a G; nucleotide 9213 is an A; nucleotide 9326 is a T; or nucleotide 10064 is an A.

59. (Original) The method of claim 44, wherein the mutation indicative of a of PKD1 associated disorder comprises a nucleotide sequence substantially identical to SEQ ID NO:1, wherein nucleotide 3336 is deleted; nucleotide 3707 is an A; nucleotide 5168 is a T; nucleotide 6078 is an A; nucleotide 6089 is a T; nucleotide 6326 is a T; nucleotides 7205 to 7211 are deleted; nucleotide 7415 is a T; nucleotide 7433 is a T; nucleotide 7883 is a T; or nucleotides 8159 to 8160 are deleted; or wherein a GCG nucleotide sequence is inserted between nucleotides 7535 and 7536.

60. (Currently amended) A method of diagnosing a PKD1-associated disorder in a subject, the method comprising:

amplifying a portion of a PKD1 polynucleotide in a nucleic acid sample from a subject with ~~at least a first primer pair~~ the set of primer pairs of claim 7 to obtain a first amplification product, ~~wherein said first primer pair is a primer pair of claim 7;~~

amplifying the first amplification product with at least a second primer pair to obtain a nested amplification product, wherein the second primer pair is suitable for performing nested amplification of the first amplification product; and

determining whether the nested amplification product has a mutation associated with a PKD1-associated disorder,

wherein the presence of a mutation associated with a PKD1-associated disorder is indicative of a PKD1-associated disorder, thereby diagnosing a PKD1-associated disorder in the subject.

61. (Original) The method of claim 60, wherein the method is performed in a high throughput format using a plurality of nucleic acid samples.

62. (Currently amended) A method of detecting the presence of a mutant PKD1 polynucleotide in a sample, the method comprising:

contacting a sample suspected of containing a mutant PKD1 polynucleotide with a polynucleotide of claim 20 under highly stringent conditions that allow the polynucleotide to selectively hybridize with a mutant PKD1 polynucleotide; and

detecting selective hybridization of the polynucleotide and a mutant PKD1 polynucleotide, thereby detecting the presence of a mutant PKD1 polynucleotide sequence in the sample.

63. (Currently amended) A kit for detecting the presence or absence of a mutation in a polycystic kidney disease-associated protein-1 (PKD1) gene, the kit comprising the set of primers of claim 1 ~~a primer, said primer comprising a 5' region and adjacent 3' region,~~
~~said 5' region comprising a nucleotide sequence that selectively hybridizes to a PKD1 gene sequence and, optionally, to a PKD1 gene homolog sequence, and~~
~~said 3' region comprising a nucleotide sequence that selectively hybridizes to a PKD1 gene sequence, and not to a PKD1 gene homolog sequence,~~
~~provided the primer does not consist of a sequence as set forth in SEQ ID NO:11, SEQ ID NO:18, SEQ ID NO:52, or SEQ ID NO:60.~~

64. (Cancelled).

65. (Currently amended) A kit for detecting the presence or absence of a mutation in a polycystic kidney disease-associated protein-1 (PKD1) gene, the kit comprising the set of primer pairs of claim 7 ~~a primer pair, said primer pair comprising a forward primer and a reverse primer,~~
~~wherein the primer pair selectively hybridize to SEQ ID NO:1 and can amplify a portion of SEQ ID NO:1 comprising about nucleotides 2043 to 4290; nucleotides 17907 to 22489; nucleotides 22218 to 26363; nucleotides 26246 to 30615; nucleotides 30606 to 33957; nucleotides 36819 to 37140; nucleotides 37329 to 41258; nucleotides 41508 to 47320; or a combination thereof.~~

66. (Previously presented) A kit for detecting the presence or absence of a mutation in a polycystic kidney disease-associated protein-1 (PKD1) gene, the kit comprising an isolated polynucleotide, said polynucleotide comprising a contiguous sequence of at least ten nucleotides substantially identical to a nucleotide sequence of SEQ ID NO:1 or to a nucleotide sequence

complementary thereto, wherein the contiguous nucleotide sequence comprises with respect to SEQ ID NO:1, nucleotide 474, wherein nucleotide 474 is a T; nucleotide 487, wherein nucleotide 487 is an A; nucleotide 3110, wherein nucleotide 3110 is a C; a position corresponding to nucleotide 3336, wherein nucleotide 3336 is deleted; nucleotide 3707, wherein nucleotide 3707 is an A; nucleotide 4168, wherein nucleotide 4168 is a T; nucleotide 4885, wherein nucleotide 4885 is an A; nucleotide 5168, wherein nucleotide 5168 is a T; nucleotide 6058, wherein nucleotide 6058 is a T; nucleotide 6078, wherein nucleotide 6078 is an A; nucleotide 6089, wherein nucleotide 6089 is a T; nucleotide 6195, wherein nucleotide 6195 is an A; nucleotide 6326, wherein nucleotide 6326 is a T; a position corresponding to nucleotides 7205 to 7211, wherein nucleotides 7205 to 7211 are deleted; nucleotide 7376, wherein nucleotide 7376 is a C; a nucleotide sequence corresponding to nucleotides 7535 to 7536, wherein a GCG nucleotide sequence is inserted between nucleotides 7535 and 7536; nucleotide 7415, wherein nucleotide 7415 is a T; nucleotide 7433, wherein nucleotide 7433 is a T; nucleotide 7696, wherein nucleotide 7696 is a T; nucleotide 7883, wherein nucleotide 7883 is a T; nucleotide 8021, wherein nucleotide 8021 is an A; a nucleotide sequence corresponding to nucleotide 8159 to 8160, wherein nucleotides 8159 to 8160 are deleted; nucleotide 8298, wherein nucleotide 8298 is a G; nucleotide 9164, wherein nucleotide 9164 is a G; nucleotide 9213, wherein nucleotide 9213 is an A; nucleotide 9326, wherein nucleotide 9326 is a T; nucleotide 9367, wherein nucleotide 9367 is a T; nucleotide 10064, wherein nucleotide 10064 is an A; nucleotide 10143, wherein nucleotide 10143 is a G; nucleotide 10234, wherein nucleotide 10234 is a C; nucleotide 10255, wherein nucleotide 10255 is a T; or a combination thereof.

67. (Cancelled).

68. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 7, ~~wherein the primer pair~~ which comprises SEQ ID NOS:3 and 4.

69. (Currently amended) The ~~primer pair~~ set of primer pairs of claim 7, ~~wherein the primer pair~~ which comprises SEQ ID NOS:19 and 20.

70 and 71. (Cancelled)

72. (Currently amended) The method of claim 25, wherein the ~~primer pair~~ set of primer pairs comprises SEQ ID NOS:3 and 4.

73. (Previously presented) The method of claim 72, wherein, upon generating a PKD1-specific amplification product, the method further comprises:

contacting the PKD1-specific amplification product with at least a second primer pair comprising SEQ ID NOS:19 and 20; and

identifying the presence or absence of a mutation in the nested amplification product, thereby detecting the presence or absence of a mutation in the PKD1 polynucleotide in the sample.

74. (Currently amended) The method of claim 44, wherein the ~~at least one primer pair~~ set of primer pairs comprises SEQ ID NOS:3 and 4.

75. (Previously presented) The method of claim 53, wherein the second primer pair comprises SEQ ID NOS:19 and 20.